WORLD DUCHENNE ORGANIZATION

Sprint #4 Bridging the silos between Clinical Research, eHealth & Digital Therapeutics
“The rise of patient-centric applications”

''Challenges and Opportunities for Patient Organizations in the Digital Era: The Duchenne Example''

Dimitrios Athanasiou
European Medicines Agency PDCO Member
World Duchenne Organization Board Member
European Patient Forum Board Member
Email : dimitrios.athanasiou@worldduchenne.org

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I have no real or apparent relevant financial relationships to disclose

DOI

www.ema.europa.eu/docs/en_GB/document_library/contacts/athanasioud_DI.pdf

The Rare Disease Example

The Rare Disease Example

Since 2000

2067 Orphan designations

171 Orphan designations included in authorised indication

153 Authorised OMPs

61 To be used in children

4 Removed from the market

43 Marketed, but no longer "orphans"

To date

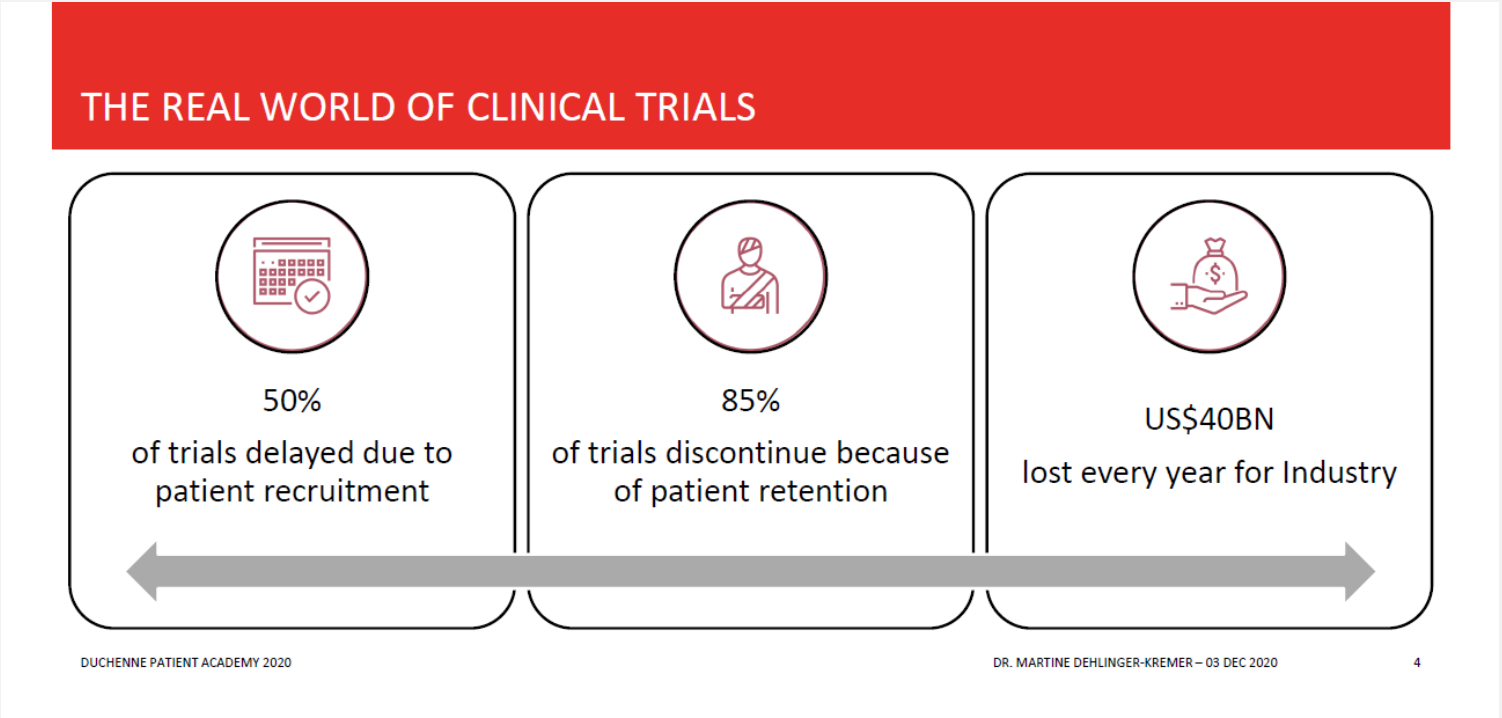
169

Products with a marketing authorisation and an orphan status in the European Union

Paediatric Unmet need

Rare Diseases, Paediatric Cancers, Neonatals……we are failing them

Why ?



Credit : Chris Barbalis@cbarbalis

Why ?

Fragmentation of Efforts and Data

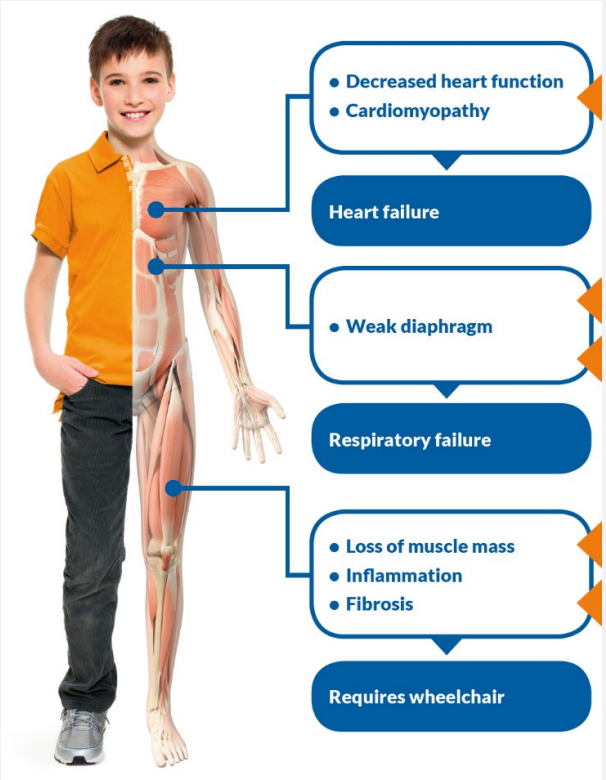
- Diagnosis, Care & Management
- Basic Research
- Clinical trial design
- Drug Development Pipeline
- Regulatory Pathway
- Access Pathway
- Policy

Credit : Chris Barbalis@cbarbalis

The Duchenne Example

Credit : Chris Barbalis@cbarbalis

Duchenne & Becker MD



DMD gene: one of the **longest** (2.3 mb)

MUTATIONS +++

Evolving Natural History

The History

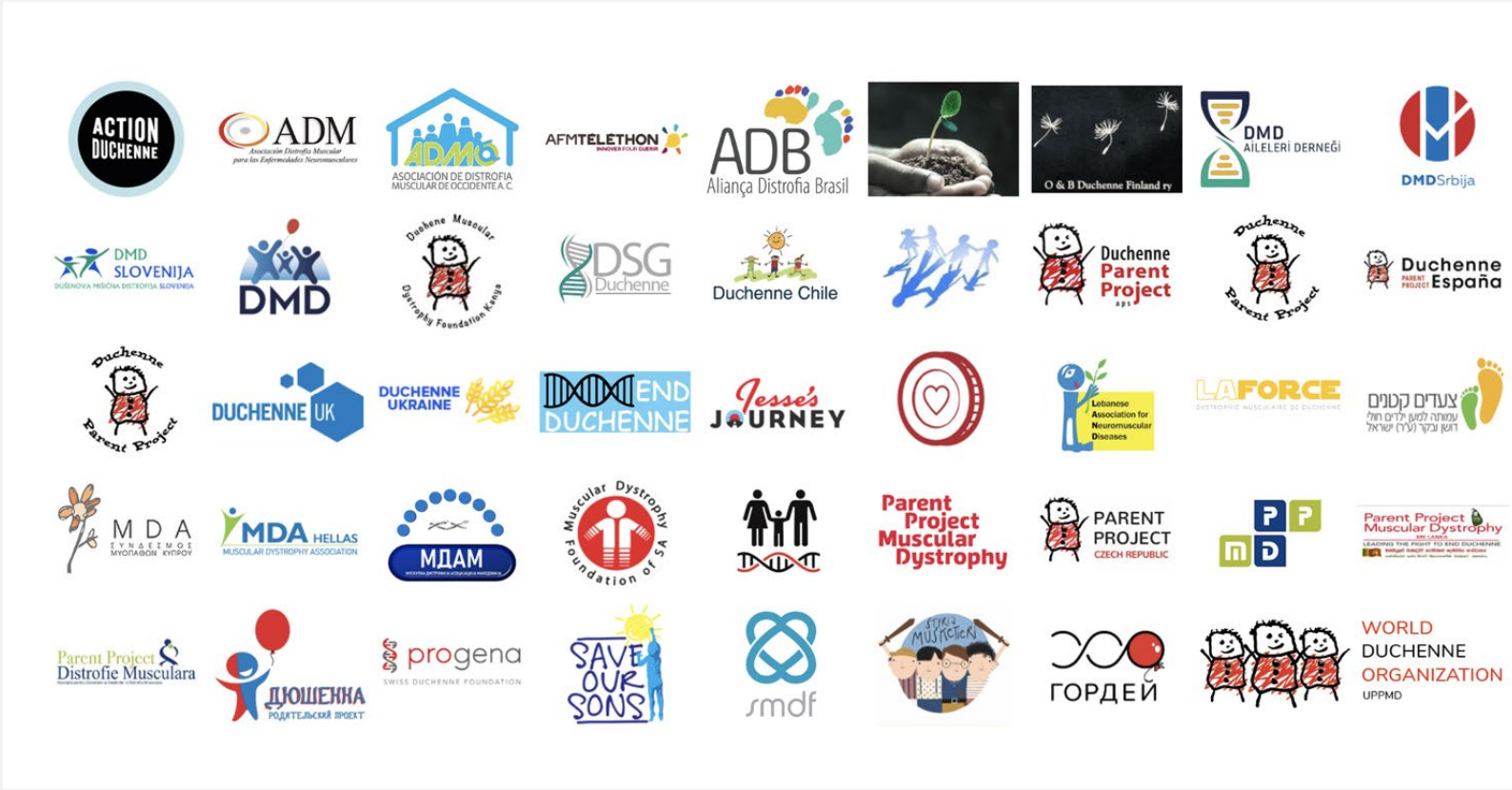
- **1860s,** was first described by the French neurologist Guillaume Benjamin Amand Duchenne
- **1950s** the first NMD Patient Organizations were founded both in Europe and US
- **1980s** new Duchenne and Becker Patient Organizations have been created all over the world
- **2000s** they have evolved to a vibrant global network of advocacy groups that shaped a lot of the rare disease advocacy into the new millennium

- **At the dawn of 2020s** the Duchenne Community is not any more the grassroots collective of desperate parents that were trying to save their children

Although it keeps its **strong drive, can-do mentality and passion** it more closely resembles a **well-oiled advocacy machine** that finances and shapes medical innovation and ATMs, co designs and influences Regulation and Policy in a global level.

Credit : Chris Barbalis@cbarbalis

45 MEMBER PATIENT ORGANIZATIONS FROM 38 COUNTRIES



The Results

The DMD community is funding 80M $ from Research to Care and Medicines Development :

- Disease Prognostic Models
- Preclinical research support
- In Silico Development A.I. and Machine learning
- Animal models
- Virus Development
- ATM research like Gene and Cell Therapies,
- Gene editing and Exon skipping technologies
- Biotech and Spin-off seed funding
- Regulatory Guidelines for DMD Developers
- Clinical Trails Simulation Tools development
- Duchenne Platform Trials development
- New outcome measures development
- PROMs and PROs development and validation
- Care Guidelines development
- HTA and Reimbursement Models development
- Exoskeletons and Supportive Digital Applications

Credit : Chris Barbalis@cbarbalis

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DUCHENNE FAIR DATA DECLARATION

1. Patient derived or provided data are not owned by those who collect them, and their reuse should be primarily controlled by the donors of these data. Researchers, charities, companies and health professionals are custodians.

2. To enable the optimal reuse of data, the data needs to be Findable, Accessible, Interoperable and Reusable (i.e. FAIR) by medical professionals, patients and in particular also by machines.

3. The optimal reuse of data should be supported at all levels, by professionals and custodians (allowfederated learning on the data upon request, give the data to the donor in FAIR format when asked), care professionals (capture data at the source in FAIR format wherever possible), analytics environments (adapt to FAIR data) and regulators (demand FAIR data throughout and optimally use them in the regulatory process). There is a need to educate all stakeholders about the FAIR principles and their importance ('FAIR Aware')

4. Optimal care should be taken to restrict the need to reveal the actual identity of individuals associated with certain data, and to protect privacy with all possible means, but we realize privacy is subordinate in many cases to fast-tracking of better solutions for the diseases we suffer from.

5. Therefore, the right to allow identification of the individual associated with certain data should also be placed in the hands of that individual or a chosen trusted party.

6. Techniques and tools should be developed to enable optimal co-investigation by researchers, medical and health care professionals, charities, companies, patients and machines to form a 'social health machine' aimed at better solutions and care.

7. Regulators should optimally enable fast-tracking of key interventions and involve citizen and machine participation in that process to the largest possible extent.

8. The field should actively discourage publishing of health-related information exclusively in classical narrative journals. These are very difficult to access and understand by both informed lay people and by machines. Instead, data and information should be published in a way that makes it more readily reusable by others than a small inner circle.

9. Funding agencies should have good data stewardship following the FAIR principles included in their grant conditions.

10. The role of health insurance institutions/companies should also be made clear: they should publicly state that having full access to real world data, even when these are re-identifiable for them to 'their' clients, will not be abused, such as for example increase premiums based on genetic predisposition.

11. Health insurance institutions/companies and governments should join forces and support (also financially) the development of trusted environments where real world citizen data can be maximally reused for the betterment of health care and the massive saving of costs to keep optimal healthcare affordable for all.

WORLD DUCHENNE ORGANIZATION

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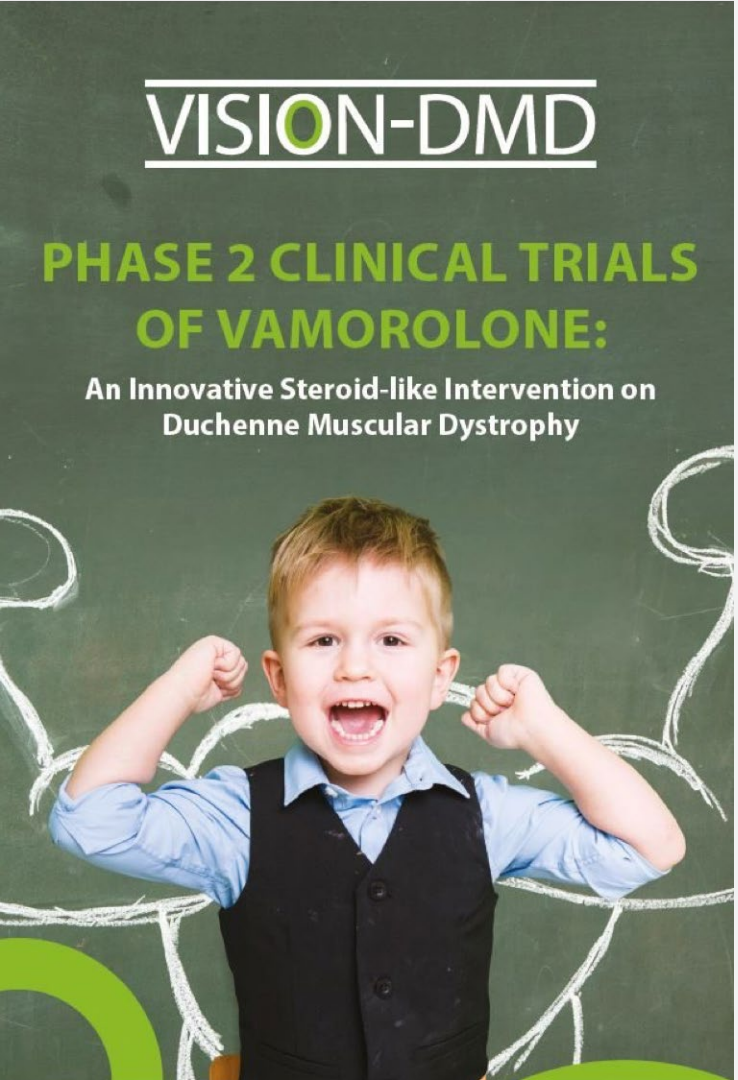
Founded in 2007 with a strong focus on:

- **Advocacy**
- **Policy**
- **Regulatory work**

Global network of member patient organizations
WDO is member of EURORDIS and European Patient's Forum (EPF)
WDO Board Members are involved in FDA and in EMA
Eligible member of the European Medicines Agency (EMA)



VISION-DMD

PROJECT TO ASSESS VAMOROLONE SAFETY

- ➔ Leverage EU funding to deliver a DMD clinical trial
- ➔ Assess Vamorolone safety and efficacy
- ➔ How to use venture philanthropy and pool funding to collect resources for setting up a clinical trial
- ➔ Return of patient data to participating families

https://vision-dmd.info

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 667078.

BIND PROJECT

LEARNING & BEHAVIOUR ISSUES IN DMD/BMD

- Brain comorbidities are a burden to DMD/BMD families
- Aims to perform the largest deep brain phenotyping effort for dystrophinopathy performed to date
- Consortium tries to address these challenges and collect data to deal with the problem
- WDO will collect feedback and experience from community to identify possible comorbidities, their prevalence and severity
- WDO allows outreach to more than 1.000 patients

https://bindproject.eu

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 847826.

BIND

SHARE4RARE

PATIENT-LED RARE DISEASE RESEARCH

- ➔ Project aims to connect patients via symptom similarity, instead of their rare condition
- ➔ Reach ultra-rare neuromuscular conditions and run pilot
- ➔ Open for patients and patient organizations, can be used to identify new patients and connect to other NMD organizations
- ➔ JWMDRC and WDO lead questionnaire on how caring for a child with a rare disease affects educational and career opportunities

https://share4rare.org

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 780262.

TRIALS@HOME

CLINICAL TRIALS DELIVERED AT HOME

- ➔ Exploring opportunities for remote decentralized clinical trials (RDCTs) in Rare Diseases if applicable
- ➔ Assess technologies applicable for remote monitoring
- ➔ Minimize the patient burden of patients and families participating in CTs
- ➔ Opportunity to learn from other stakeholders (tech, regulatory, drug development)
- ➔ Facilitating dialogue with relevant stakeholders and developing learnings and networks for RDCTs in DMD

https://trialsathome.com

This project has received funding from the Innovative Medicines Initiative 2 Joint Undertaking under grant agreement No 831458. This Joint Undertaking receives support from the European Union's Horizon 2020 research and innovation programme and EFPIA.

EURO-NMD Registry Hub

European Reference Network for neuromuscular diseases

Key Facts

EURO-NMD Registry Hub
Start date: 01-05-2020
End date: 01-05-2023
www.registry.ern-euro-nmd.eu
info@registry.ern-euro-nmd.eu

Objective

Building a registry hub for all neuromuscular diseases, including undiagnosed patients, and connect with existing ones.

Partners

- Assistance Publique Hopitaux de Paris
- Universitaetsklinikum Freiburg
- Stichting Katholieke Universiteit
- World Duchenne Organization
- Duchenne Data Foundation
- Association Institut de Myologie
- Association Francaise Contre Les Myopathies

Patient centered and interoperable registry hub for Rare Neuromuscular Diseases

EURO-NMD, ERN for Rare Neuromuscular Diseases, spans 14 European countries, with 61 reference centres that oversee more than 100,000 patients. Core objectives are the implementation of clinical practice guidelines and the definition and monitoring of core indicators of guideline conforming management, treatment quality and patient health outcomes.

Patient registries are key instruments for the ERN to be able to deliver its objectives. A recognised challenge for rare diseases is the heterogeneity of legacy data sets and the multiplicity of existing registries. EURO-NMD health care providers and patient organizations are currently active in more than 120, mostly disease specific and patient run registries. While the existing registries are collecting important information, none of them is used by all EURO-NMD centres and there is no unified NMD or NMD Disease Specific Registry in EU.

The general objective is to build a registry hub for all neuromuscular diseases, including undiagnosed patients, and connect with the existing ones. The EURO-NMD Registry Hub will use internationally agreed, state of the art concepts such as being built with a system that will collect standardized common data elements, defined by the Joint Research Center (JRC).

It will be registered in the JRC's meta-registry platform ERDRI, will allow for the generation of a Privacy Preservation Record Link (PPLR) through the EUPID system, it will use internationally accepted ontologies (HPO) and ORPHA codes for codification of the diseases. Development of a registry hub that will allow linking and extraction of data from different sources. Thus, the data collected through the registry hub will be Findable, Accessible, Interoperable, and Reusable (FAIR).

The registry hub will offer the unique opportunity to the fragmented NMD communities and their HCPs, Centers, Patients and Patient Organizations to be able to communicate between them and exchange knowledge, experience and news.

This project has received funding from the 3rd EU Health Programme under grant agreement No 947598

EURO-NMD REGISTRY HUB

RARE NMD DATA HUB

- ➔ Improving quality and equity of healthcare
- ➔ Enabling the exchange of knowledge through teaching and training
- ➔ Facilitating translational research
- ➔ Focus on strengthening the collaboration amongst the 61 HCPs so that, when new members are proposed and endorsed, their integration is swift with minimum impact on the ERN's activities.

https://registry.ern-euro-nmd.eu/

This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement number 947598.

DUCHENNE

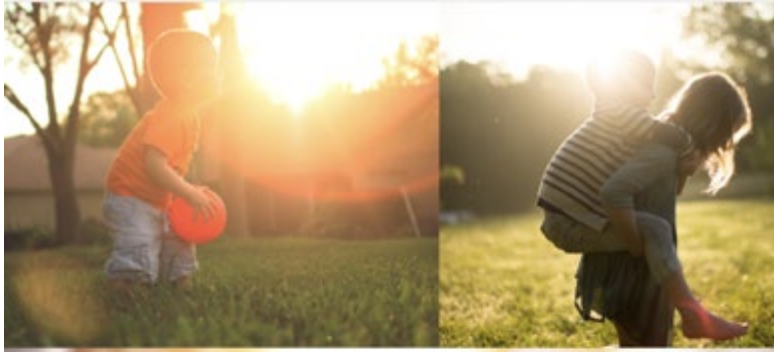
DATA FOUNDATION

Founded in 2015 with a strong focus on:

- **Data**
- **Research**
- **Training**

➔ Patient-led foundation focusing on data collection and harmonization to facilitate and optimize DMD/BMD research & development.

➔ DDF provides the framework and structure enabling research, knowledge exchange and education in the patient community and with other stakeholders.



DUCHENNE

DATA FOUNDATION

DUCHENNE MAP

SOCIAL DUCHENNE

DUCHENNE

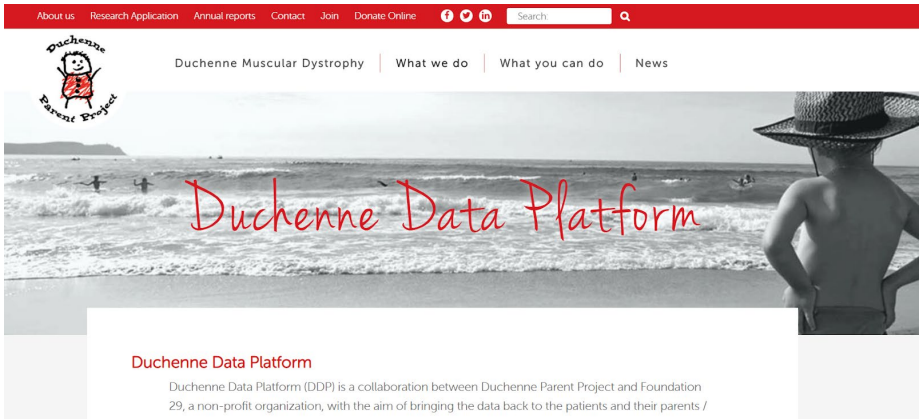
COMMUNITY ADVISORY BOARD

DUCHENNE

DATA REPOSITORY

DUCHENNE

PATIENT ACADEMY



BIND

Brain Involvement in Dystrophinopathies

EURO-NMD Registry Hub

European Reference Network for neuromuscular diseases

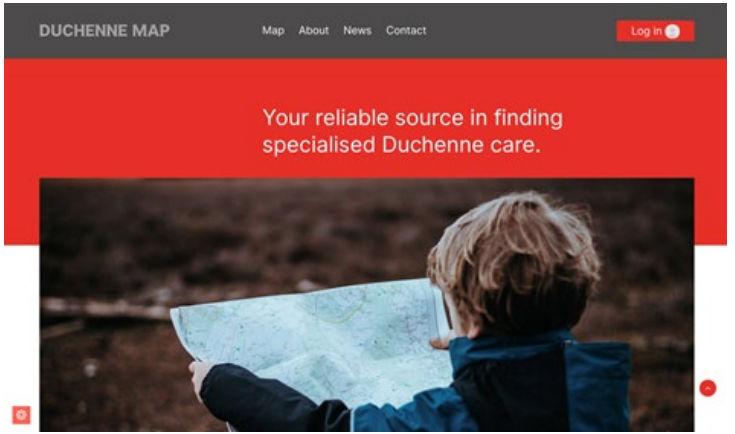
DUCHENNE MAP

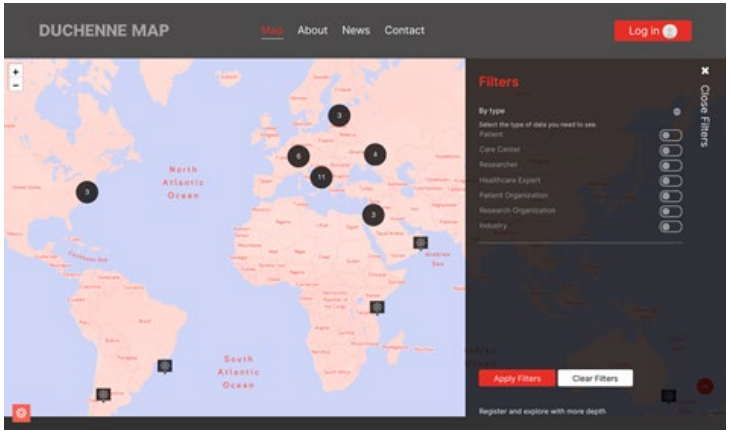
Interactive tool to centralise DMD information

Hub to connect global DMD stakeholders

Collect and analyse data to improve the lives of DMD families

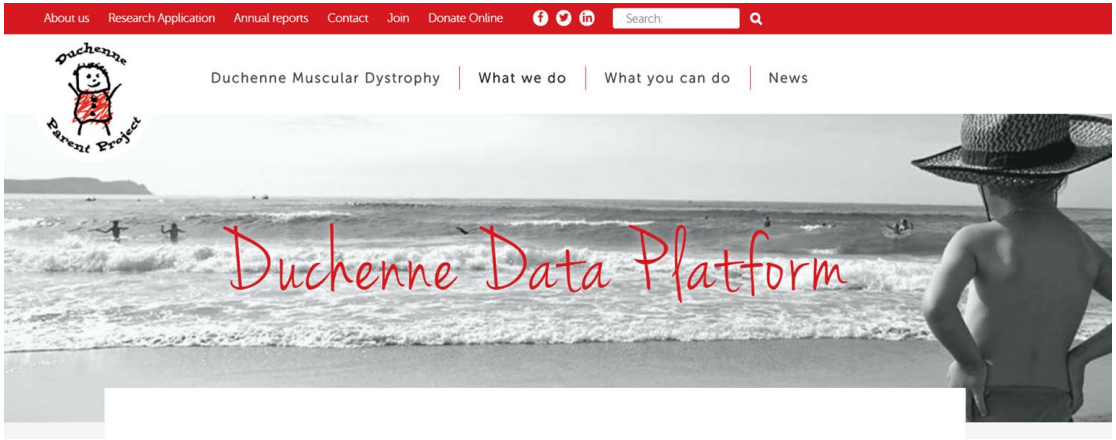
www.duchennemap.org





Duchenne Data Platform

A patient-led registry



www.duchennedataplatform.org

About:

- Built in 2019
- Patients health data in one place: ‘Lockers’
- PROMs are collected
- GDPR-compliant
- Patient Informed Consent in place
- Double authentication
- Access data through wearables
- Promoted internationally
- Undertaking a FAIRification process (Completion date December 2021)

FAIR contact: Nawel van Lin

nawel@duchennedatafoundation.org

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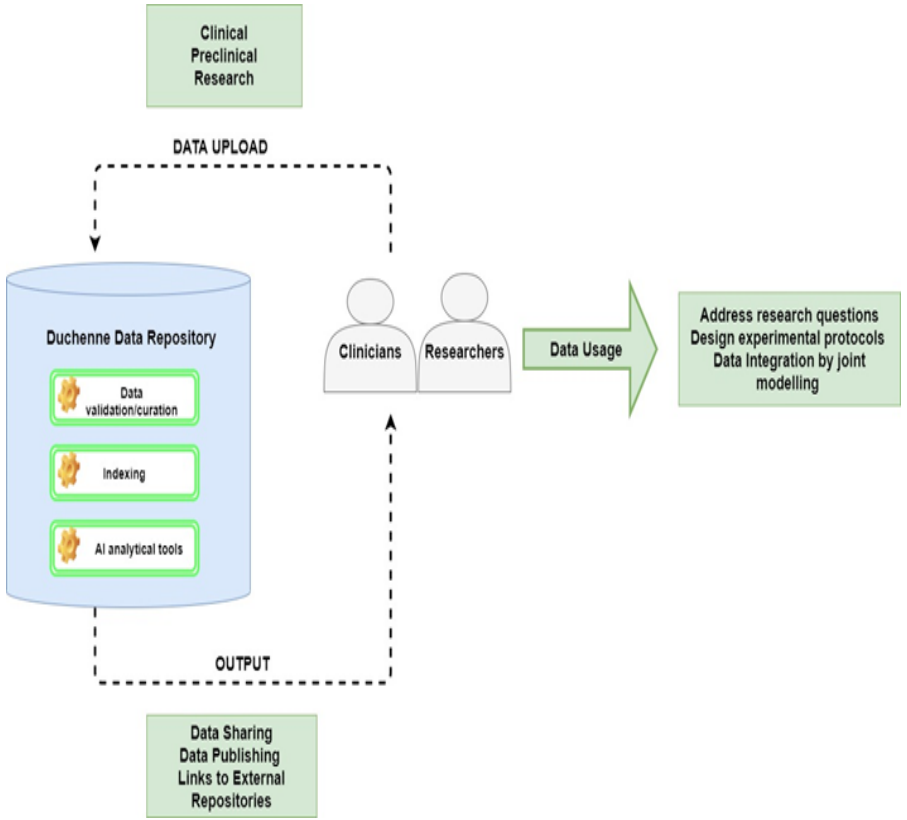
DATA REPOSITORY

Aims

- Create a ***unified environment*** for data from **different sources**
- Facilitate the ***sharing*** and ***re-use*** of data and information
- **Improve care** and **accelerate the discovery** of effective treatments for DMD/BMD patients



BETWEEN HYPE AND HOPE

- Still the pathway is marked with many success and failures.
- The boys live longer, have a better a life but still lose the fight with Duchenne.
- Balancing between Hype and Hope the community still fights to keep the boys alive while keeping the Rare Disease in the centre of R&D and Regulatory discussions.

THE CHALLENGES

- Many unknows in patient management and care
- Many unknows in disease trajectory and evolving SoC
- Clinical Trials Design (ex. Master Protocol, End Points, Biomarkers etc)
- Data Issues and in Silo attitudes (# of Registries)
- Collection of Reliable Data and Long Term Follow Up (DMD Post Marketing Registry)
- Pipeline issues , with many failing products but also Feasibility issues - # of drugs

Access to innovative therapies and good care

WHAT THE FUTURE HOLDS?

Create collectively a blueprint to optimize the pathway from Research to Access;

- To develop technologies and tools together with the patients
- To collect patient-centric, relevant, reliable and interoperable data
- To drive evidence/data based advocacy
- To accelerate science/data driven drug development and care pathways
- To foster cooperation and collaboration between all stakeholders for the use of the data and these tools

WHAT THE FUTURE HOLDS?

A lot of work ☺

We will build together an inclusive value framework where each stakeholder can contribute and where we can all co-create to improve paediatric research and care for the benefit of our children and our health systems

THANK YOU!

ATHANASIOU DIMITRIOS, BA, MBA, World Duchenne Organization / UPPMD EPF Board Member, EMA PDCO Member